



CCBE1 gene

collagen and calcium binding EGF domains 1

Normal Function

The *CCBE1* gene provides instructions for making a protein that is found in the lattice of proteins and other molecules outside the cell (extracellular matrix). The *CCBE1* protein is involved in the formation of the lymphatic system, which consists of a network of vessels that transport lymph fluid and immune cells throughout the body. Specifically, the *CCBE1* protein helps guide maturation (differentiation) and movement (migration) of immature cells called lymphangioblasts that will eventually form the lining (epithelium) of lymphatic vessels.

Health Conditions Related to Genetic Changes

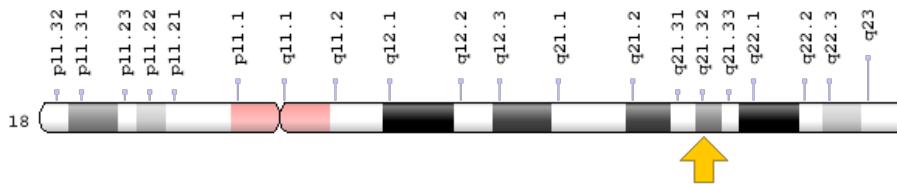
Hennekam syndrome

At least 13 mutations in the *CCBE1* gene have been found to cause Hennekam syndrome, an inherited disorder resulting from malformation of the lymphatic system. Most *CCBE1* gene mutations change single protein building blocks (amino acids) in the *CCBE1* protein leading to a change in the three-dimensional shape of the protein. The abnormal protein cannot play its role in the formation of the lymphatic vessel epithelium. A poorly formed lymphatic system leads to lymphatic vessels that are abnormally expanded (lymphangiectasia) and are prone to break open (rupture), puffiness or swelling caused by a buildup of fluid (lymphedema), and other features of Hennekam syndrome. *CCBE1* gene mutations account for about 25 percent of all cases of Hennekam syndrome.

Chromosomal Location

Cytogenetic Location: 18q21.32, which is the long (q) arm of chromosome 18 at position 21.32

Molecular Location: base pairs 59,430,939 to 59,698,181 on chromosome 18 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- collagen and calcium-binding EGF domain-containing protein 1
- FLJ30681
- full of fluid protein homolog
- KIAA1983

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Human Lymphoid Organs (figure)
<https://www.ncbi.nlm.nih.gov/books/NBK26921/figure/A4423/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CCBE1%5BTIAB%5D%29+OR+%28EGF+domains+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- COLLAGEN AND CALCIUM-BINDING EGF DOMAIN-CONTAINING PROTEIN 1
<http://omim.org/entry/612753>

Research Resources

- **Atlas of Genetics and Cytogenetics in Oncology and Haematology**
http://atlasgeneticsoncology.org/Genes/GC_CCBE1.html
- **ClinVar**
<https://www.ncbi.nlm.nih.gov/clinvar?term=CCBE1%5Bgene%5D>
- **HGNC Gene Symbol Report**
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=29426
- **NCBI Gene**
<https://www.ncbi.nlm.nih.gov/gene/147372>
- **UniProt**
<http://www.uniprot.org/uniprot/Q6UXH8>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/CCBE1>

Reviewed: July 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services